

Table 7. TTR Variants and Their Phenotypes

Mutation	Exon	Codon Change	Phenotype	Geographic Focus
Gly6Ser	Exon 2	GGT-AGT	non-amyloid, FEH	Caucasian
Cys10Arg		TGT-CGT	Heart, eye, PN	USA
Leu12Pro		CTG-CCG	LM, liver	UK
Met13Ile		ATG-ATC	non-amyloid	Germany
Asp18Glu		GAT-GAA	PN	South America, USA
Asp18Gly		GAT-GGT	LM	Hungary
Asp18Asn		GAT-AAT	Heart	USA
Val20Ile		GTC-ATC	Heart, CTS	Germany, USA
Ser23Asn		AGT-AAT	Heart, PN, eye	USA
Pro24Ser		CCT-TCT	Heart, CTS, PN	USA
Ala25Ser		GCC-TCC	Heart, CTS, PN	USA
Ala25Thr		GCC-ACC	LM, PN	Japan
Val28Met		GTG-ATG	PN, AN	Portugal
Val30Met		GTG-ATG	PN, AN, eye, LM	Portugal, Japan, Sweden, USA
Val30Ala		GTG-GCG	Heart, AN	USA
Val30Leu		GTG-CTG	PN, heart	Japan
Val30Gly		GTG-GGG	LM, eye	USA
Val32Ala		GTG-GCG	PN	Israel
Phe33Ile		TTC-ATC	PN, eye	Israel
Phe33Leu		TTC-CTC	PN, heart	USA
Phe33Val		TTC-GTC	PN	UK, Japan, China
Phe33Cys		TTC-TGC	CTS, heart, eye, kidney	USA
Arg34Thr		AGA-ACA	PN, heart	Italy
Arg34Gly		AGA-GGA	Eye	UK
Lys35Asn		AAG-AAC	PN, AN, heart	France
Lys35Thr		AAG-ACG	Eye	USA
Ala36Pro		GCT-CCT	Eye, CTS	USA
Asp38Ala		GAT-GCT	PN, heart, lung	Japan
Trp41Leu		TGG-TTG	Eye, PN	USA
Glu42Gly		GAG-GGG	PN, AN, heart	Japan, USA, Russia
Glu42Asp		GAG-GAT	Heart	France
Phe44Ser		TTT-TCT	PN, AN, heart	USA
Ala45Thr		GCC-ACC	Heart	USA
Ala45Asp		GCC-GAC	Heart, PN	USA
Ala45Ser		GCC-TCC	Heart	Sweden
Gly47Arg		GGG-CGG/AGG	PN, AN	Japan
Gly47Ala		GGG-GCG	Heart, AN	Italy, France
Gly47Val		GGG-GTG	CTS, PN, AN, heart	Sri Lanka
Gly47Glu		GGG-GAG	Heart, PN, AN	Turkey, USA, Germany
Thr49Ala	Exon 3	ACC-GCC	Heart, CTS	France, Italy
Thr49Ile		ACC-ATC	PN, heart	Japan, Spain
Thr49Pro		ACC-CCC	Heart, PN	USA
Ser50Arg		AGT-AGG	AN, PN	Japan, France/Italy, USA
Ser50Ile		AGT-ATT	Heart, PN, AN	Japan
Glu51Gly		GAG-GGG	Heart	USA
Ser52Pro		TCT-CCT	PN, AN, Heart, kidney	UK
Gly53Glu		GGA-GAA	LM, heart	Basque, Sweden
Gly53Ala		GGA-GCA	LM, heart	UK
Glu54Gly		GAG-GGG	PN, AN, eye	UK

Glu54Lys		GAG-AAG	PN, AN, heart, eye	Japan
Glu54Leu		GAG-CTG	not described	UK
Leu55Pro		CTG-CCG	Heart, AN, eye	USA, Taiwan
Leu55Arg		CTG-CGG	LM	Germany
Leu55Gln		CTG-CAG	Eye, PN	USA
Leu55Glu		CTG-CAG	Heart, PN, AN	Sweden
His56Arg		CAT-CGT	Heart	USA
Gly57Arg		GGG-AGG	Heart	Sweden
Leu58His		CTC-CAC	CTS, heart	USA (MD) (FAP II)
Leu58Arg		CTC-CGC	CTS, AN, eye	Japan
Thr59Lys		ACA-AAA	Heart, PN, AN	Italy, USA (Chinese)
Thr60Ala		ACT-GCT	Heart, CTS	USA (Appalachian)
Glu61Lys		GAG-AAG	PN	Japan
Glu61Gly		GAG-GGG	Heart, PN	USA
Phe64Leu		TTT-CTT/TTG	PN, CTS, heart	USA, Italy
Phe64Ser		TTT-TCT	LM, PN, eye	Canada, UK
Ile68Leu		ATA-TTA	Heart, PN	Germany
Tyr69His		TAC-CAC	Eye, LM	Canada, USA
Tyr69Ile		TAC-ATC	Heart, CTS, AN	Japan
Lys70Asn		AAA-AAC	Eye, CTS, PN	USA
Val71Ala		GTG-GCG	PN, Eye, CTS	France, Spain
Ile73Val		ATA-GTA	PN, AN	Bangladesh
Asp74His		GAC-CAC	non-amyloid	Germany
Ser77Tyr		TCT-TAT	Heart, Kidney, PN	USA (IL, TX), France
Ser77Phe		TCT-TTT	PN, AN, heart	France
Tyr78Phe		TAC-TTC	PN, CTS, skin	France
Ala81Thr		GCA-ACA	Heart	USA
Ala81Val		GCA-GTA	Heart	UK
Ile84Ser		ATC-AGC	Heart, CTS, eye	USA (IN), Hungary
Ile84Asn		ATC-AAC	Heart, eye	USA
Ile84Thr		ATC-ACC	Heart, PN	Germany, UK
His88Arg		CAT-CGT	Heart	Sweden
Glu89Gln		GAG-CAG	PN, heart	Italy
Glu89Lys		GAG-AAG	PN, heart	USA
His90Asn		CAT-AAT	non-amyloid	Germany, Portugal
His90Asp		CAT-GAT	Heart	UK
Ala91Ser		GCA-TCA	PN, CTS, heart	France
Glu92Lys		GAG-AAG	Heart	Japan
Val94Ala		GTA-GCA	Heart, PN, AN, kidney	Germany, USA
Ala97Gly	Exon 4	GCC-GGC	Heart, PN	Japan
Ala97Ser		GCC-TCC	PN, heart	Taiwan, USA
Gly101Ser		GGC-AGC	non-amyloid	Japan
Pro102Arg		CCC-CGC	non-amyloid	Germany
Arg103Ser		CGC-AGC	Heart	USA
Arg104Cys		CGC-TGC	non-amyloid	USA
Arg104His		CGC-CAC	non-amyloid, FEH	Japan, USA(Chinese)
Ile107Val		ATT-GTT	Heart, CTS, PN	USA
Ile107Met		ATT-ATG	PN, heart	Germany
Ile107Phe		ATT-TTT	PN, AN	UK
Ala109Thr		GCC-ACC	non-amyloid, FEH	Portugal
Ala109Val		GCC-GTC	non-amyloid, FEH	USA

Ala109Ser		GCC-TCC	PN, AN	Japan
Leu111Met		CTG-ATG	Heart	Denmark
Ser112Ile		AGC-ATC	PN, heart	Italy
Tyr114Cys		TAC-TGC	PN, AN, eye, LM	Japan, USA
Tyr114His		TAC-CAC	CTS, skin	Japan
Tyr116Ser		TAT-TCT	PN, CTS, AN	France
Thr119Met		ACG-ATG	non-amyloid, FEH	Portugal, USA
Ala120Ser		GCT-TCT	AN, Heart, PN	Afro-Caribbean
Val122Ile		GTC-ATC	Heart	USA
del Val122		GTC-	Heart, PN	USA (Ecuador), Spain
Val122Ala		GTC-GCC	Heart, eye, PN	USA
Pro125Ser		CCC-TCC	non-amyloidogenic	Italy

AN, autonomic neuropathy; CTS, carpal tunnel syndrome; FEH, familial euthyroid hypertyroxinemia;
LM leptomeningeal PN, peripheral neuropathy

This table is derived from Connors et al, 2003 and Benson et al, 2007